

## Additional file 1

**Table S1.** Distribution of OHIP-14 questionnaire

OHIP domains	Descriptive of each question	Total LDS (N = 33) % subject yes (N)
Functional Limitation	OHIP 1 - Had trouble pronouncing words	36.36 (12)
	OHIP 2 - Felt sense of taste has worsened	
Pain	OHIP 3 - Had painful aching in the mouth	57.58 (19)
	OHIP 4 - Found it uncomfortable to eat any foods	
Discomfort	OHIP 5 - Have been self-conscious	51.52 (17)
	OHIP 6 - Felt tense	
Physical Disability	OHIP 7 - Had an unsatisfactory diet	30.30 (10)
	OHIP 8 - Had to interrupt meals	
Psychological Disability	OHIP 9 - Found it difficult to relax	42.42 (14)
	OHIP 10 - Have been a bit embarrassed	
Social Disability	OHIP 11 - Have been irritable with other people	21.21 (7)
	OHIP 12 - Had difficulty doing usual jobs	
Handicap	OHIP 13 - Felt life in general was less satisfying	12.12 (4)
	OHIP 14 - Have been totally unable to function	

**Table S2:** Comparison of OHIP-14 health subdomains between three different age groups in the LDS cohort

Dimension	Age			P- value
	Childhood	Adolescence	Adult	
	OHIP-14 Mean $\pm$ SD			
Functional Limitation	1.56 $\pm$ 1.6	0.58 $\pm$ 1.2	0.58 $\pm$ 0.9	0.14
Pain	1.22 $\pm$ 1.5	1.75 $\pm$ 1.5	1.25 $\pm$ 1.6	0.66
Discomfort	0.33 $\pm$ 0.7	1.75 $\pm$ 1.8	1.58 $\pm$ 1.6	0.09
Physical Disability	0.77 $\pm$ 1.6	0.42 $\pm$ 0.9	1.5 $\pm$ 1.9	0.23
Psychological Disability	0.67 $\pm$ 1.4	0.92 $\pm$ 1.1	1.17 $\pm$ 1.1	0.64
Social Disability	0.78 $\pm$ 0.9	0.33 $\pm$ 0.8	0.75 $\pm$ 1.2	0.57
Handicap	0.22 $\pm$ 0.7	0.08 $\pm$ 0.3	0.58 $\pm$ 1.4	0.43

Childhood: includes subjects whose age was 11 years old (y.o.) or less

Adolescence: includes subjects whose age was greater than 11 y.o. and less than 18 y.o.

Adult: includes subjects whose age was greater than 18 y.o.

**Statistical analyses:** ANOVA test was employed.

**Table S3:** Comparison of OHIP-14 health subdomains between males and females in LDS cohort

Dimension	Gender		<i>P</i> - value
	Female	Male	
	OHIP-14 Mean $\pm$ SD		
Functional Limitation	0.53 $\pm$ 1.0	1.18 $\pm$ 1.4	0.13
Pain	1.35 $\pm$ 1.6	1.50 $\pm$ 1.5	0.79
Discomfort	1.41 $\pm$ 1.5	1.12 $\pm$ 1.7	0.61
Physical Disability	1.00 $\pm$ 1.7	0.81 $\pm$ 1.5	0.73
Psychological Disability	0.88 $\pm$ 0.9	1.13 $\pm$ 1.7	0.61
Social Disability	0.59 $\pm$ 1.1	0.44 $\pm$ 0.8	0.66
Handicap	0.47 $\pm$ 1.3	0.13 $\pm$ 0.3	0.30

**Statistical analyses:** Mann-Whitney U test was employed.

**Table S4:** Comparison of OHIP-14 health subdomains between types of LDS mutations

Dimension	Type of LDS Mutations			<i>P</i> - value
	LDS1	LDS2	LDS3-5	
	OHIP-14 Mean $\pm$ SD			
Functional Limitation	1.13 $\pm$ 1.5	0.58 $\pm$ 1.0	0.67 $\pm$ 1.2	0.50
Pain	1.27 $\pm$ 1.4	1.42 $\pm$ 1.6	1.83 $\pm$ 1.8	0.75
Discomfort	1.40 $\pm$ 1.6	1.17 $\pm$ 1.7	2.00 $\pm$ 2.3	0.65
Physical Disability	0.67 $\pm$ 1.4	1.17 $\pm$ 1.9	1.00 $\pm$ 1.3	0.71
Psychological Disability	1.20 $\pm$ 1.6	0.83 $\pm$ 1.1	0.83 $\pm$ 1.0	0.74
Social Disability	0.40 $\pm$ 0.8	0.83 $\pm$ 1.3	0.17 $\pm$ 0.4	0.33
Handicap	0.07 $\pm$ 0.3	0.67 $\pm$ 0.1.5	0.17 $\pm$ 0.4	0.25

LDS1: LDS patients with TGFBR1 mutation

LDS2: LDS patients with TGFBR2 mutation

LDS3-5: LDS patients with either SMAD3, TGFB2, or TGFB3 mutation

**Statistical analyses:** ANOVA test was employed.

**Table S5:** Frequency of oral manifestations for each age group in the LDS cohort

Oral manifestations	Age		
	Childhood (N = 10)	Adolescence (N = 12)	Adult (N = 11)
	% (N)		
<i>Malocclusion</i>	90.0% (9)	100.0% (12)	100.0% (11)
<i>Abnormal soft and hard palate</i>	40.0% (4)	50.0% (6)	81.8% (9)
<i>Gingivitis</i>	30.0% (3)	83.3% (10)	63.6% (7)
<i>Hypersensitivity</i>	90.0% (9)	100.0% (12)	72.7% (8)
<i>TMJ abnormality</i>	20.0% (2)	33.3% (4)	72.7% (8)
<i>Self-rated poor-to-fair oral health status</i>	20.0% (2)	33.3% (4)	45.5% (5)
<i>Cumulation of oral manifestations (<math>\geq 4</math>)</i>	20.0% (2)	75.0% (9)	81.8% (9)

**Table S6:** Influence of food allergy on OHIP-14 (OHRQoL) in LDS patients

Variables	% (n)	OHIP-14	P-value
		Mean $\pm$ SD	
Food Allergy			0.68
Yes	30.3 % (10)	5.60 $\pm$ 1.7	
No	69.7% (23)	6.61 $\pm$ 1.4	

**Statistical analyses:** Mann-Whitney U test was employed.